



## What is it?

### Points To Remember About Osteogenesis Imperfecta (OI)

- OI is a genetic disease that causes bones to weaken and break easily.
- There are several types of OI. The type of OI you have affects the symptoms you have.
- Symptoms of OI range mild to severe. All people with OI have weak bones that break easily.
- OI is usually managed by a team of doctors.
- There are several treatments for OI, including medicines and surgery. Treatments may help prevent or control symptoms of OI.
- There are steps you can take to stay healthy and help prevent broken bones.
- There is no way to prevent OI.

Osteogenesis imperfecta (OI) is a disease that affects your bones. OI is also called brittle bone disease. If you have OI, you have bones that are weak and break easily.

## Who gets it?

OI is a genetic disease, which means it runs in certain families. About 20,000 to 50,000 people in the United States have OI. OI is present at birth and is a lifelong disease.

## What are the types?

There are eight types of OI. The type of OI you have affects how mild or severe the symptoms

of the disease is. Type 1 is the mildest and most common form of OI. Type 2 is the most severe form of OI. Other types of OI have symptoms that fall between Type 1 and Type 2.

## **What are the symptoms?**

All people with OI have weak bones that break easily. People with OI often have other symptoms, such as:

- Malformed bones.
- Short, small body.
- Skin the bruises easily.
- Loose joints.
- Weak muscles.
- Whites of the eyes that look blue, purple, or gray.
- A face shaped like a triangle.
- A rib cage shaped like a barrel.
- A curved spine.
- Brittle teeth.
- Hearing loss, often starting in 20s or 30s.
- Breathing problems.

OI symptoms vary from person to person. They can range from mild to severe. For example, some people with OI may have a few broken bones over their lifetime. Others may have hundreds of broken bones in their lifetime, including broken bones that occur before birth.

## **What causes it?**

OI is caused by an abnormal gene. Genes carry information that determine which features are passed to you from your parents.

People with OI have a defect in one of the genes that helps make collagen. Collagen is a substance in the body that makes bones strong. People with OI do not have enough collagen. Or, the collagen does not work properly. This causes weak bones that break easily.

Most people with OI inherit this gene from one parent. Others inherit it from both parents. Parents do not have to have OI to pass on the gene that causes it.

Sometimes, neither parent passes on the gene. Instead, the gene stops working properly on its own before the child is born.

## **Is there a test?**

There is no single test to detect OI. Doctors look at several factors to diagnose OI. These include:

- Family history.
- Medical history.
- Results from a physical exam.
- X-rays.

OI can be diagnosed through a skin biopsy. A skin biopsy is a surgical procedure in which a doctor removes a small piece of skin. Then, a doctor looks at it under a microscope to see whether there are any collagen abnormalities.

Doctors can also diagnose OI through a blood test that detects the abnormal genes that causes OI.

## How is it treated?

The goal of treatment is to prevent or control symptoms of OI.

### Medicines

- **Bone strengthening medicines:** Your doctor may prescribe bone strengthening medicines to slow bone loss and reduce the frequency and seriousness of broken bones.
- **Pain medicines:** Your doctor may prescribe pain medicines to treat pain caused by broken bones.

### Broken Bone Care

Your doctor may cast, splint, or brace a broken bone to help it heal correctly.

### Surgery

Your doctor may recommend surgery for many reasons:

- To fix a broken bone.
- To support or correct that are curved or bowed.
- To support the spine.

Many children with OI have surgery in which a metal rod is placed into a bone. This is called rodding surgery. Rodding surgery is performed to support the bone and prevent the bone from breaking.

### Mobility Aids

Your doctor may recommend a walker, cane, crutches, or wheelchair to reduce injuries.

## Physical or Occupational Therapy

You may benefit from physical or occupational therapy, which can help you:

- Build muscle strength, which may help prevent broken bones.
- Learn how to avoid injuries.
- Safely perform activities of daily living.
- Recover from broken bones.

## Dental Care

Some people with OI have brittle teeth that chip or crack easily. You may require special dental care.

## Who treats it?

If you have OI, you will likely require a health care team that includes made up of several doctors and other health care providers. These include:

- Internists, who diagnose and treats adults
- Pediatricians, who diagnose and treats children
- Orthopaedists, who treat and perform surgery for bone and joint diseases
- Occupational therapists, who teach how to safely perform activities of daily living.
- Physical therapists, who teach ways to build muscle strength and prevent broken bones. Physical therapists may also help you recover from broken bones.
- Nutritionists, who teach how to use diet to improve overall health and stay at a healthy weight.

## Living With It

There are steps you can take to stay healthy and help prevent broken bones.

- Follow a nutritious diet.
- Exercise as much as possible. Regular physical activity can help strengthen muscles and bones. Swimming and water therapy are common choices for people with OI because exercising in water has little risk of broken bones. Talk with your doctor before beginning any new exercises.
- Keep a healthy weight. Being overweight increases the risk for many health problems, such as diabetes and heart disease. Extra weight also adds stress to the bones, which is especially unhealthy for people with OI.
- Don't smoke. Smoking causes cancer, heart disease, and other serious health conditions. Smoking can also weaken bones.
- Do not drink a lot of alcohol or caffeine. They may weaken your bones.

- Do not take steroid medicines. They can weaken bones.

## For More Info

### **U.S. Food and Drug Administration**

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

[Drugs@FDA](https://www.accessdata.fda.gov/scripts/cder/daf) at <https://www.accessdata.fda.gov/scripts/cder/daf> [Drugs@FDA](https://www.accessdata.fda.gov/scripts/cder/daf) is a searchable catalog of FDA-approved drug products.

### **Centers for Disease Control and Prevention, National Center for Health Statistics**

Website: <https://www.cdc.gov/nchs>

### **NIH Osteoporosis and Related Bone Diseases ~ National Resource Center**

Website: <https://www.bones.nih.gov>

### **Osteogenesis Imperfecta Foundation**

Website: <https://www.oif.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at [NIAMSInfo@mail.nih.gov](mailto:NIAMSInfo@mail.nih.gov).

- [Asian Language Health Information](#)
- [Spanish Language Health Information](#)

### **Join a Clinical Trial**

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### **Related Information**

[Osteogénesis Imperfecta: Esenciales: hojas informativas de fácil lectura](#)

### **View/Download/Order Publications**

[Osteogenesis Imperfecta, Easy-to-Read Fast Facts](#)

[Osteogenesis Imperfecta Overview](#)

[OI Issues: Maintaining Health During the Adult Years](#)